

SHORT REPORT

Hereditary chin trembling or hereditary chin myoclonus?

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Abstract

Hereditary chin trembling is a rare autosomal dominant disease often considered as an "essential tremor variant". The clinical and neurophysiological data obtained in a new white family lead to the suggestion that this abnormal involuntary movement is a focal variant of hereditary essential myoclonus.

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Hereditary chin trembling¹ is a rare autosomal dominant condition described in the literature as "geniospasm", "facial spasm", "Ein streng dominant erbliches kinnmuskelszittern", "familial trembling of the chin", "hereditary quivering of the chin", and "trembling chin". In 1993¹ up to 21 affected families had been reported. The male:female ratio was 1.3:1. There were 20 white families (United States and central Europe) and one black family. We present a new white family (figure 1) which gives us the opportunity to discuss the nature of the abnormal involuntary movement.

The index case (IV-13) was a 35 year old sales representative who was examined because of abnormal involuntary movement of the chin. This had been present since birth, constantly

during childhood, and then episodically, lasting a few minutes. It appeared electively in stressful situations such as playing videogames, pinball, or in working hours when meeting customers. He had been free of symptoms for two years but the abnormal involuntary movement reappeared a few months before examination after buying a computer for work and pleasure. It was described as a tremulous movement of the chin but he said that once he had noted that the abnormal involuntary movement had spread to the upper lip. During some periods with severe stress, the abnormal involuntary movement was so great that it was present during sleep and sometimes woke him up.

He was unable to initiate or suppress it. On examination a quick, up and down, tremulous bilateral movement was sometimes spontaneously seen at the tip of his chin for a few minutes. Whether this abnormal involuntary movement was present or not, there was neither postural hand tremor, head tremor, voice tremor, or palatal myoclonus, and a general neurological examination was negative. He wanted to be treated because he found the abnormal involuntary movement socially disabling in his professional activities even though he knew that it was benign, many members of his family having had the same problem.

His brother (IV-12) had had the same abnormal involuntary movement since birth. It was less severe and without social or

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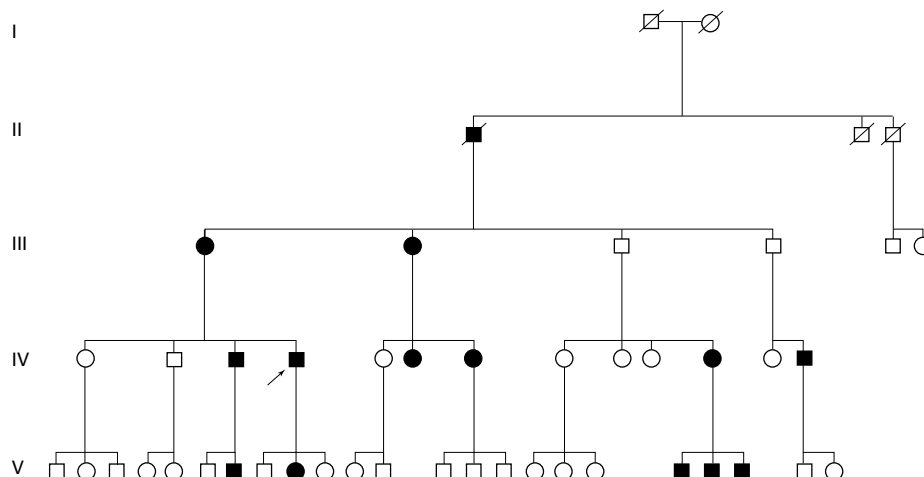


Figure 1 Pedigree of family. Affected members in black (♂=proband).

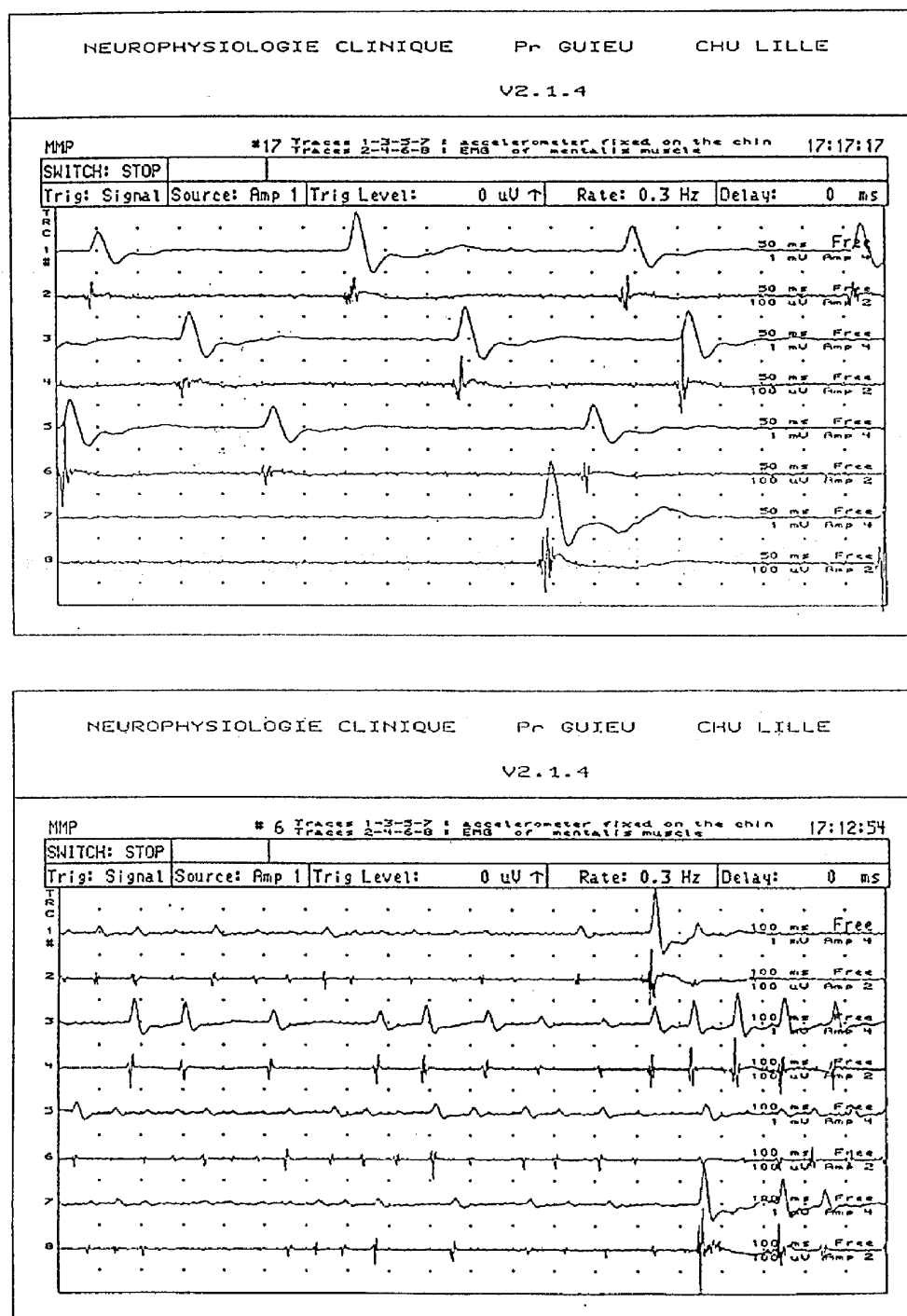


Figure 2 Surface EMG activity of mentalis muscle (traces 2, 4, 6, 8) and "tremorogram" (traces 1, 3, 5, 7) obtained from an accelerometer placed on the chin. During playing activities, there are brief bursts of EMG, pseudorhythmic or irregular, without sinusoidal oscillations.

professional consequences. Two female first cousins (IV-8 and IV-9) presented the abnormal involuntary movement with the same characteristics. A 30 year old male first cousin (IV-1) and three female first cousins (IV-6: 34 years old; IV-8: 43 years old ; IV-9: 27 years old) had had very slight abnormal involuntary movement from birth until they were a few months old. An 11 year old boy (V-20) was socially disabled because under some circumstances the abnormal involuntary movement could be so intense that speaking was impaired.

A 4 year old girl (V-22) was the daughter of the proband. The trembling chin had been present very early in life; it had been seen by her father a few minutes after birth, in the delivery room. According to her father the abnormal involuntary movement could be seen when she was sleeping. Three young boys (V-6, V-7, and V-8) had the same abnormal involuntary movement but very rarely. The mother of the proband (III-6) was 62 years old. She also had had the chin tremor when an infant but it progressively disappeared with time. It had not been

clinically seen for many years but she said that she felt shiverings when stressed. A 65 year old woman (III-5) had had chin tremor when a child but it was rapidly only seen in stressful situations and had not been noticed for 10 years. Two men (III-3 and III-4) were said never to have had chin tremor but they both had a child with this abnormal involuntary movement. No patient's symptoms were improved by alcohol intake. No member of this family, with or without an abnormal involuntary movement of the chin, had other abnormal involuntary movements.

Electrophysiological studies were carried out on the proband. The EMG activity was obtained from mentalis muscle with two surface electrodes placed 2 cm apart, filtered (20 Hz-2 kHz), and amplified. An accelerometer was attached to the chin. The signal was fed to the EMG device and to a PC to obtain spectral analysis. Several 10 second recordings were made with a sampling rate of 100 Hz. When appropriate, different activation manoeuvres were used to enhance the movement disorder, such as playing with his daughter. Figure 2 shows the results.

The peak frequency of chin "tremor" varied widely between 5.7 and 10.3 Hz. Intermittently, brief (10-25 ms duration) bursts of activity were seen, especially during stressful and playing activities. Sometimes the bursts discharged in a rhythmic fashion and had the appearance of a clinical tremor; but most often the frequency of discharge was irregular.

Simultaneous recording of video, EEG, and EMG of the mentalis muscle was made with digital equipment (Deltamed, Paris). EEG activity was obtained from 19 surface electrodes placed on the scalp according to the international 10-20 system and low pass filtered at 256 Hz with a time constant of one second. The EEG signals were back averaged off line with respect to the EMG bursts.

Standard EEG showed no anomaly. Back averaging the EEG before the jerks failed to disclose any potential.

Sensory evoked potentials were measured using an eight channel Nicolet Pathfinder II, with the subject lying supine in a quiet dark room. Median nerves were stimulated at the wrist with surface electrodes, using 0.1 ms constant current square pulses with an intensity of 1.2 above motor threshold at a rate of 3.3 Hz. Recordings were obtained from Erb's point, cervical (C6), C'3, C'4, F'3, and F'4, with an earlobe reference contralateral to the stimulation, and also from the abductor pollicis brevis muscle ipsilateral to the stimulation. Trigeminal evoked potentials were obtained by stimulating the skin of the lips with surface electrodes, using 0.1 ms constant current square pulses with an intensity of three times subjective threshold, at 2.7 Hz. Cortical responses were recorded from an electrode placed contralateral to the stimulation side on the midpoint of a line joining the external auditory meatus and a point 1 cm posterior to Cz, with an Fz and earlobe reference. For each nerve, two series of 500 stimulations were averaged.

All cortical responses were of normal latency and amplitude. There was no C-reflex, even after muscular activation (for upper limbs).

Discussion

These patients share the features of those recorded by Danek,¹ but there are some other characteristics. (1) In one case the abnormal involuntary movement of the chin was sufficiently severe to impede speech, as recently reported.² (2) Two subjects can be considered as free of symptoms but were obligatory gene carriers suggesting incomplete penetrance. (3) In some cases "the first episodes appear shortly after birth"¹ but to our knowledge a patient with "chin trembling" in the very first minutes of life has never been reported. This family gives us the opportunity to discuss the nature of the abnormal involuntary movement.

Do our patients present an "essential tremor variant"?³ Some features are not in favour of such a hypothesis. Essential tremor rarely begins in infancy and childhood,^{4,5} contrary to what occurred with our patients and those already reported.^{1,2} It is a slowly progressive disorder unlike the case with our patients, whose abnormal involuntary movement progressively and sometimes very quickly disappeared. Essential tremor can concern some body parts such as the chin, but only in patients presenting with the classic, often severe symptoms,⁶ which have never been noticed in our patients. The only index case of the first family reported by Soland *et al*² had "a minimal postural tremor of the outstretched hands". It must also be emphasised that no case of essential tremor has been found in the families reported until now. In action tremor burst duration is 50 to 250 ms.⁷ On a clinical basis the "tremor of the smile"⁸ can easily be excluded.

Are the words "tremor" or "trembling" appropriate? A tremor is usually described as: "rhythmical involuntary oscillatory movement of a body part",⁹ "periodic movement about an axis",¹⁰ "any involuntary approximately rhythmic, roughly sinusoidal movement",⁴ and "involuntary oscillation of a body part produced by alternating or synchronous contractions of reciprocally innervated antagonistic muscles".¹¹ All these definitions emphasise the notions of oscillation and rhythmicity which suggest either alternating or synchronous activity in antagonist muscles. In our patients as well as in others the abnormal involuntary movement is not really rhythmic. That it is due to the activity of only one muscle is not important; some tremors are related to the activity of only one antigravitic muscle.¹⁰ The abnormal involuntary movement of the chin is seen in stressful or emotional situations but cannot be related to rest, posture, and action (which characterise the different types of tremor)¹⁰ and contrary to tremor⁵ it can be seen during sleep (two; personal case). The neurophysiological records show that the movement was not sinusoidal and that there was an interval between each movement. So we think that the chin abnormal involuntary movement cannot be considered as "a tremor". When isolated, "site

specific tremors³ were traditionally considered as focal manifestations of essential tremor. However, "focal tremor could be viewed as a manifestation of a covert dystonia".¹² Neither our patients, nor the members of their families, nor those reported in the literature presented with symptoms suggesting dystonia as found in this condition.¹³

Some authors considered the abnormal involuntary movement to be myokymia and proposed the term "familial myokymia of the chin".¹⁴ Myokymiae are in fact very different from what was found in our patients. They are unilateral and localised but concern several muscles and are rhythmic and regular without influence of stress or emotion. They are more rapid¹⁴ and their neurophysiological aspects are different.¹⁵ Also, myorhythmia can be excluded because it is slower and usually involves the face, the eyes, and the limbs.¹⁶

So, finally we think that our patients presented with "hereditary chin myoclonus". Indeed there are many factors in favour of such a diagnosis: (1) the abnormal involuntary movement is related to quick jerks of the mentalis muscle; (2) the burst duration is brief (10–25 ms); (3) the wave form of the abnormal involuntary movement is very similar to that of rhythmical (or pseudorhythmical) myoclonus, which resembles a series of sharp spikes; there are also pauses between the individual jerks.¹⁰ A cortical origin can reasonably be excluded, as sensory evoked potentials (by median and trigeminal nerve stimulation) are not enhanced and EEG back averaging is negative.¹⁷ The clinical data are not in favour of reticular myoclonus.¹⁸ Therefore this myoclonus seems to be subcortical in origin.

The "hereditary chin myoclonus" is different from palatal and ocular myoclonus and from facial action myoclonus.¹⁵

It could be considered as a focal variant of hereditary essential myoclonus¹⁹ but with some characteristics: its presence during sleep, its spontaneously favourable evolution, sometimes disappearing quickly, and the lack of response to alcohol.²⁰

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